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## Autosomal Dominant Polycystic Kidney Disease

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## Autosomal Dominant Polycystic Kidney Disease

Jonathan S. Maltzman, MD, PhD

### INTRODUCTION

Autosomal dominant polycystic kidney disease (ADPKD) is one of the most common hereditary disorders, occurring in 1 in 400 to 1000 live births.<sup>1</sup> Approximately 500,000 persons in the United States have ADPKD. It is the third most common entity leading to end-stage renal disease (ESRD) in the United States, accounting for 8% to 10% of cases of ESRD. Most affected individuals will progress to ESRD by the seventh decade of life.

ADPKD is only one of a number of cystic disorders, both acquired and hereditary, that affect the kidney. It is characterized by cysts distributed throughout both kidneys, although the disease is not limited to the kidney and abnormalities are commonly found in the liver, heart, and cerebral vasculature. ADPKD is inherited as an autosomal dominant trait that has nearly complete penetrance, but there is phenotypic variability even within families. Although great strides have been made in recent years in understanding both the genetics and the pathophysiology of ADPKD, much remains unknown. This manual reviews some of the genetic and pathogenic mechanisms of ADPKD and describes the management of this common renal disease.

### CLASSIFICATION OF RENAL CYSTIC DISEASE

#### CASE PATIENT PRESENTATION

A 35-year-old man with a history of hypertension is referred to a nephrologist for evaluation of mild renal insufficiency and bilateral renal cysts.

#### HISTORY

The patient had presented 2 weeks earlier to a local emergency room with flank pain and gross hematuria. Diagnostic evaluation with computed tomography (CT) scanning was done to evaluate for nephrolithiasis and revealed multiple cysts throughout both kidneys and no

evidence of nephrolithiasis (**Figure 1**). The patient's serum creatinine level was 1.3 mg/dL. He was given nonsteroidal anti-inflammatory drugs (NSAIDs) for the pain. The gross hematuria subsided within 3 days, and the pain has since improved. Additional history obtained at the time of initial consultation revealed that the patient's father had "something wrong with his kidneys" and died of a cerebrovascular accident at age 45 years. The patient has 2 younger siblings (aged 25 and 32 years) with no known renal disease and 2 children (aged 3 and 6 years) with no known medical problems.

The patient is currently taking benazepril 10 mg daily, which he has been on for 7 years. He states that he is not taking any other medications. He denies using NSAIDs in the past week. Review of systems is significant for occasional back pain; he denies experiencing fevers, dysuria, headaches, or visual changes.

#### PHYSICAL EXAMINATION

Physical examination reveals a well-nourished, well-developed man with a blood pressure of 150/90 mm Hg and heart rate of 70 bpm. Heart and lung examinations are normal. Abdominal examination reveals mild hepatomegaly, and the kidneys are palpable bilaterally. There is no peripheral edema. Neurologic examination is normal.

- What is the differential diagnosis of cystic disease in this patient?

#### DIFFERENTIAL DIAGNOSIS

Patients are commonly referred to a nephrologist following demonstration of renal cysts on imaging studies. There is a broad differential diagnosis for cystic diseases of the kidneys (**Table 1**). Some disorders are entirely benign, while others have a tendency to progress to ESRD and the need for renal replacement therapy. It is important to differentiate between acquired cystic disease and hereditary forms and to understand medical conditions associated with each. This review focuses on ADPKD and only briefly describes other conditions that may be confused with ADPKD.